



# AMYLOIDOSIS PROGRAM

A team approach to diagnose and care for patients with amyloidosis



**Allegheny**  
Health Network

Cardiovascular Institute



# The AHN Amyloidosis Program supports patients



**Comprehensive treatment team of experts** in many subspecialties who coordinate individualized treatment plans.



**Regional support center** to build a community of patients, families, and caregivers; connect them with local resources; and raise public awareness.



**Leading research center** that gives our patients access to the latest clinical trials and investigational therapies as we learn more about this family of diseases.



# What is amyloidosis?

Amyloidosis (am-uh-loy-DO-sis or amyloid) is a rare disease that occurs when the body incorrectly deposits misfolded (defective) proteins in various organs, disrupting normal organ function.

More than 30 separate proteins have been found in different people with amyloidosis. In general, different proteins tend to build up in different organs. Amyloidosis that affects the heart is almost always due to one of two proteins that was inappropriately deposited there.

amyloidosis can affect nearly every organ in the body. In addition to the heart, it can affect the kidneys, nervous system, digestive tract, liver, muscles, and skin. As the disease progresses, amyloidosis can lead to life-threatening organ failure.

In addition to treating the organ dysfunction, treatment of amyloidosis also works to decrease the production of defective proteins to prevent them from building up in the organs in the first place.

Except in rare cases, there is no cure for amyloidosis. However, treatments help make patients feel significantly better and live substantially longer.

## Possible amyloidosis symptoms

While these symptoms are not specific to amyloidosis, they may raise the need for additional testing.

- Heart failure with preserved ejection fraction (congestive heart failure due to a heart stiffened by protein buildup).
- Unexplained left ventricular hypertrophy (thickening of the heart muscle).
- Cardiac arrhythmias (irregular heartbeats that may be too fast or too slow).
- Orthostatic hypotension (dizziness or fainting with changes in position).
- Spinal stenosis (low back pain).
- Carpal tunnel syndrome (numbness, tingling, or weakness in the hand).
- Unexplained peripheral neuropathy (weakness, numbness, or pain in the hands or feet).
- Renal failure with proteinuria (abnormal kidney function).
- Macroglossia (an unusually large tongue).



## Types of amyloidosis

The two main proteins that deposit in the heart include antibody fragments called “light chains” and a vitamin A/thyroid hormone transporter called transthyretin (TTR).

**Light chain amyloidosis** (AL amyloidosis, formerly known as primary amyloidosis) occurs when excess antibody fragments called light chains are overproduced by the bone marrow in a cancer-like process and then circulate through the blood and build up in various organs. AL amyloidosis can affect the heart, kidneys, skin, nerves, and liver.

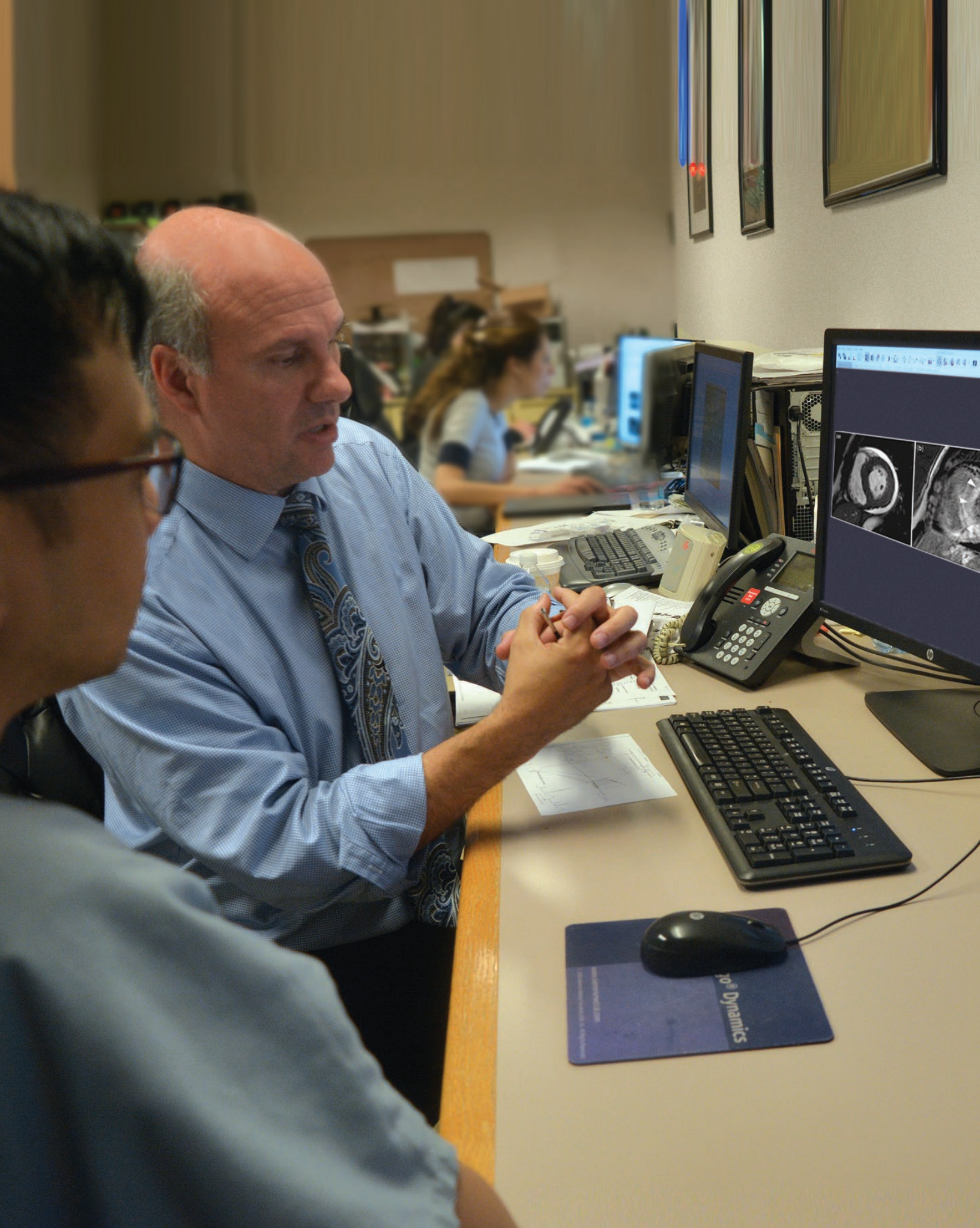
Affected patients may have amyloidosis alone or in association with other plasma cell dyscrasias (multiple myeloma).

**Transthyretin amyloidosis** (ATTR amyloidosis) occurs when excess, defective transporter proteins called TTR are produced by the liver and circulate through the blood to various organs. There are two types of ATTR:

- A protein that is defective due to a mutation the patient was born with (mutant or hereditary TTR, or hTTR).
- A protein that is defective due to mistakes in protein creation, which occur more frequently as the patient ages (wild-type TTR or wtTTR, formerly known as “senile amyloidosis”).

There are several other types of amyloidosis, including those specific to patients on dialysis (primarily affecting their joints) and those with inflammatory diseases like rheumatoid arthritis (primarily affecting their kidneys).





## How is amyloidosis diagnosed?

Because there are many nonspecific signs and symptoms of amyloidosis, the average patient will see several doctors over many years before they receive a true diagnosis. After diagnosis, subtyping (identifying the culprit protein) is critical because it affects a patient's treatment and expected course (prognosis).

### Laboratory tests

We analyze blood and urine for abnormal circulating proteins that can suggest amyloidosis. Other laboratory tests may not show the specific proteins, but they will show organ damage where defective proteins are deposited.

### Imaging tests

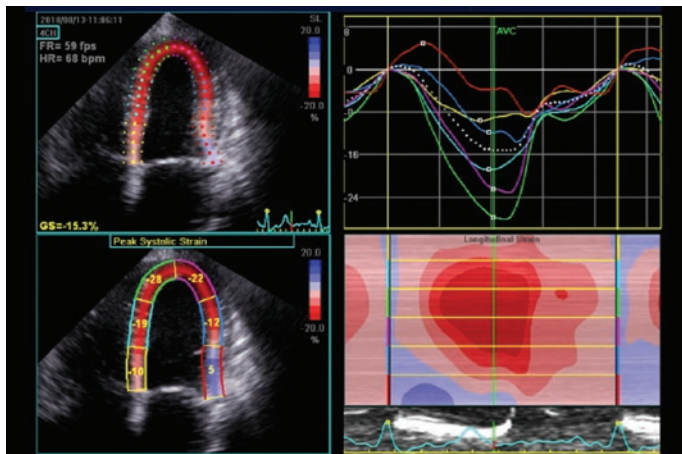
Images of the affected organs — including ultrasound (echocardiogram), MRI, and/or nuclear study (technetium pyrophosphate scan) — can help establish the extent of the disease.

### Neurologic tests

In addition to offering standard neurologic testing (EMG/NCV), the clinical neurophysiology laboratory at Allegheny General Hospital provides state-of-the-art Quantitative Sudomotor Axon Reflex Testing (QSART) and autonomic test batteries to provide a more sensitive way of diagnosing those with amyloidosis.

### Biopsy

In some cases, to firmly establish an amyloidosis diagnosis, we must obtain a small piece of tissue from an affected organ and study it under a microscope. This is critical to confirm protein deposits and identify the specific protein that is involved. The biopsy may be performed on the bone marrow, on the heart through an outpatient heart catheterization, or on the skin and soft tissue.



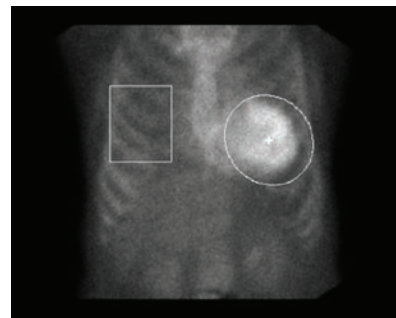
Transthoracic echocardiogram with strain imaging showing relative apical sparing consistent with cardiac amyloidosis.

## Treatments

Treatment for amyloidosis aims to:

- Prevent further protein accumulation.
- Remove already-deposited defective proteins.
- Manage impaired organ function.

For example, in patients with symptomatic cardiac amyloid, therapy tries to slow down the buildup of new protein in the heart, remove the proteins already there, and treat the heart failure that has resulted. In extreme cases, cardiac transplant may even be considered.



Technetium pyrophosphate scan showing significant uptake in the heart, strongly suggestive of TTR cardiac amyloidosis.

Clinical research has led to advances that are making this an exciting time in the treatment of TTR amyloidosis, both hereditary and wild-type.

**Treating AL amyloidosis:** Because the bone marrow overproduces antibody light chains in a cancer-like process, we use chemotherapy to destroy those light-chain producing “factories.” Sometimes a bone marrow transplant is considered.

**Treating TTR amyloidosis:** New medicines, now under review, prevent the defective TTR proteins from being deposited. Other therapies, in development or approved, will aim to prevent the liver from making the defective proteins in the first place.

- Regardless of which type of amyloidosis, small studies suggest that a combination of medications, including the antibiotic doxycycline, green tea extract, and ursodeoxycholic acid (Ursodiol) have been shown to help the body remove clumps of defective proteins from various organs while causing few side effects. They are therefore often used along with other treatments.
- For individuals with hereditary TTR, genetic testing and counseling can help families obtain a diagnosis and plan for the future. Importantly, just having the mutation does not mean that an individual will develop the disease, but it does mean that he or she can pass along that mutation and risk of the disease to the next generation.

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